



SEQUENCE LISTING

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(1) GENERAL INFORMATION:

- (i) APPLICANT: Gonzalez, Frank J.
Fernandez-Salguero, Pedro
- (ii) TITLE OF INVENTION: Methods and Compositions for Detecting
Dihydropyrimidine Dehydrogenase Splicing Mutations
- (iii) NUMBER OF SEQUENCES: 7
- (iv) CORRESPONDENCE ADDRESS:
(A) ADDRESSEE: Townsend and Townsend and Crew LLP
(B) STREET: Two Embarcadero Center, Eighth Floor
(C) CITY: San Francisco
(D) STATE: California
(E) COUNTRY: USA
(F) ZIP: 94111-3834
- (v) COMPUTER READABLE FORM:
(A) MEDIUM TYPE: Floppy disk
(B) COMPUTER: IBM PC compatible
(C) OPERATING SYSTEM: PC-DOS/MS-DOS
(D) SOFTWARE: PatentIn Release #1.0, Version #1.30
- (vi) CURRENT APPLICATION DATA:
(A) APPLICATION NUMBER: US 09/308,080
(B) FILING DATE: 28-OCT-1999
(C) CLASSIFICATION:
α¹⁰
- (vii) PRIOR APPLICATION DATA:
(A) APPLICATION NUMBER: US 60/013,835
(B) FILING DATE: 20-MAR-1996
- (viii) PRIOR APPLICATION DATA:
(A) APPLICATION NUMBER: WO PCT/US97/04269
(B) FILING DATE: 19-MAR-1997
- (ix) ATTORNEY/AGENT INFORMATION:
(A) NAME: Weber, Kenneth A.
(B) REGISTRATION NUMBER: 31,677
(C) REFERENCE/DOCKET NUMBER: 015280-271100US
- (x) TELECOMMUNICATION INFORMATION:
(A) TELEPHONE: (415) 576-0200
(B) TELEFAX: (415) 576-0300

(2) INFORMATION FOR SEQ ID NO:1:

- (i) SEQUENCE CHARACTERISTICS:
(A) LENGTH: 861 base pairs
(B) TYPE: nucleic acid
(C) STRANDEDNESS: single
(D) TOPOLOGY: linear
- (ii) MOLECULE TYPE: DNA (genomic)

(ix) FEATURE:

(A) NAME/KEY: CDS
 (B) LOCATION: 269..433
 (D) OTHER INFORMATION: /note= "dihydropyrimidine dehydrogenase (DPD) gene region of splicing mutant mRNA encoding amino acids 581-635"

(ix) FEATURE:

(A) NAME/KEY: mutation
 (B) LOCATION: replace(434, "a")
 (D) OTHER INFORMATION: /note= "G -> A mutation at 3' donor splice junction consensus sequence leading to loss of exon in DPD mRNA and DPD deficiency"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:1:

TGTTAATGAA GATAAATATT TTTGTTTTT CGCTGTTCTA AACCTAGGGT TACAAGAAGT 60
 AATTATCTG GAGCTAACAA ATACTTATT TTACCTTTT ATTGCAAGT AGTTTATGTT 120
 CAATTCTAAT TTAATGTATA TTAAAAATTC CTCTGCAAAT ATGTGAGGAG GGACCTCATA 180
 AAATATTGTC ATATGGAAAT GAGCAGATAA TAAAGATTAT AGCTTTCTT TGTCAAAAGG 240
 AGACTCAATA TCTTACTCT TTCATGAG GAC ATT GTG ACA AAT GTT TCC CCC 292
 Asp Ile Val Thr Asn Val Ser Pro
 1 5
 a¹⁰
 cont.
 ATA ATC ATC CGG GGA ACC ACC TCT GGC CCC ATG TAT GGC CCT GGA CAA 340
 Ile Ile Ile Arg Gly Thr Ser Gly Pro Met Tyr Gly Pro Gly Gln
 10 15 20
 AGC TCC TTT CTG AAT ATT GAG CTC ATC AGT GAG AAA ACG GCT GCA TAT 388
 Ser Ser Phe Leu Asn Ile Glu Leu Ile Ser Glu Lys Thr Ala Ala Tyr
 25 30 35 40
 TGG TGT CAA AGT GTC ACT GAA CTA AAG GCT GAC TTC CCA GAC AAC 433
 Trp Cys Gln Ser Val Thr Glu Leu Lys Ala Asp Phe Pro Asp Asn
 45 50 55
 GTAAGTGTGA TAAAAATCTA AAACAAGAGA ATTGGCATAA GTTGGTGAAT GTTTATTTAA 493
 ACATCCAATT CATAGGCTTA TAAATATTAA TGTGTATATT TTATCACACGA ATCTGCCAGT 553
 TGCTTGCTG ATGCATAGAA AGATAAAAAA GAAAGAAAAG CTCAAGAACT CATAAAAACC 613
 CACACAATGT GAAGCTCTGT TATAATGGG TGCCATGTAA GATGGAAGAA GTATCTACAT 673
 AAGCAGAAGG AAGAGAAATG AAATACTCAT TTTATTGAGT TGGCCCCAC TGTATGTGGC 733
 TGGTATTAT GAAGGTGATG ACCCAGGAAG AAATTGTAAA CTATAAACCA CTCCAAATAT 793
 AAACCCGAGG CAGAAGCAGC ATATCTCCTA TGAAGCCTGT ATTTACTCAG TGGGAAATAA 853
 TTTATTAA 861

(2) INFORMATION FOR SEQ ID NO:2:

- (i) SEQUENCE CHARACTERISTICS:
- (A) LENGTH: 55 amino acids
 - (B) TYPE: amino acid
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:2:

Asp Ile Val Thr Asn Val Ser Pro Ile Ile Ile Arg Gly Thr Thr Ser
 1 5 10 15

Gly Pro Met Tyr Gly Pro Gly Gln Ser Ser Phe Leu Asn Ile Glu Leu
 20 25 30

Ile Ser Glu Lys Thr Ala Ala Tyr Trp Cys Gln Ser Val Thr Glu Leu
 35 40 45

Lys Ala Asp Phe Pro Asp Asn
 50 55

(2) INFORMATION FOR SEQ ID NO:3:

- a¹⁰*
end
- (i) SEQUENCE CHARACTERISTICS:
- (A) LENGTH: 22 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

- (ix) FEATURE:
- (A) NAME/KEY: -
 - (B) LOCATION: 1..22
 - (D) OTHER INFORMATION: /note= "primer DELF1 (forward)"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:3:

TGCAAATATG TGAGGAGGGA CC

22

(2) INFORMATION FOR SEQ ID NO:4:

- (i) SEQUENCE CHARACTERISTICS:
- (A) LENGTH: 22 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

- (ix) FEATURE:
- (A) NAME/KEY: -
 - (B) LOCATION: 1..22
 - (D) OTHER INFORMATION: /note= "primer DELR1 (reverse)"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:4:

CAGCAAAGCA ACTGGCAGAT TC

22

(2) INFORMATION FOR SEQ ID NO:5:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 18 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 1..18
- (D) OTHER INFORMATION: /note= "primer DPD15F (forward)"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:5:

TTGTGACAAA TGTTTCCC

18

*a¹⁰
cont.*

(2) INFORMATION FOR SEQ ID NO:6:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 24 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 1..24
- (D) OTHER INFORMATION: /note= "primer DPD15R (reverse)"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:6:

AGTCAGCCTT TAGTCAGTG ACAC

24

(2) INFORMATION FOR SEQ ID NO:7:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 856 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:7:

TGTTAATGAA GATAAATATT TTTATTTTG CGCTATTCTA AACCTAGAGT TACAAGAAGT	60
AATTTATCTG GAGCTAACAA ATACTTATT TTACCTTTT ATTTGCAAGT AGTTTATGTT	120
CAATTCTAAT TTAATGTATA TTAAAAATTC CTCTGCAAAT ATGTGAGGAG GGACCTCATA	180
AAATATTGTC ATATGGAGAT GAGCAGATAA TAAAGATTAT AGCTTTCTT TGTCAAAAGG	240
AGACTCAATA TCTTACTCT TTCATCAGGA CATTGTGACA AATGTTCCC CCATAATCAT	300
CCGGGGAAACC ACCTCTGGCC CCATGTATGG CCCTGGACAA AGCTCCTTC TGAATATTGA	360
GCTCATCAGT GAGAAAACGG CTGCATATTG GTGTCAAAGT GTCACTGAAC TAAAGGCTGA	420
CTTCCCAGAC AACGTAAGTG TGATAAAAAT CTAAAACAAG AGAATTGGCA TAAGTTGGTG	480
AATGTTTATT TAAACATCCA ATTCA TAGGC TTATAAATAT TAATGTGTAT ATTTTATCAA	540
AGAATCTGCC AGTTGCTTG CTGATGCATA GAAAGATAAA AAAGAAAGAA AAGCTCAAGA	600
ACTCATAAAA ACCCACACAA TGTGAAGCTC GTTATAAATG GGTGCCATGT AAGATGGAAG	660
AAGTATCTAC ATAAGCAGAA GGAAGAGAAA TGAAATACTC AATTTATTGA GTTGGCCCC	720
ACTGTATGTG GCTGGCATTG ATGAAGGTGA TGACCCAGGA AGAAATTGTC ACCTATAAAT	780
CATCCAAATA TCCCGAGGCA GAAGCAGCAT CTCTCCTATG AAGTCTGTAT TTATTCAGC	840
GGGAAATAAT TTATTA	856

*a¹⁰
end.*